Opis choroby *

Definicja

A rare genetic neurological disorder characterized by hypotonia, delayed motor development, dyskinesia of the limbs, intellectual disability with impaired speech development, seizures, autistic features, stereotypic movements, and sleep disturbance. Onset of symptoms is in infancy. Bilateral abnormalities in the putamen on brain MRI have been reported in some patients.

Dane

Klasyfikacja
Choroba

Kod ORPHA
468620

Kod OMIM
617171

Kod ICD10
G93.8

Kod ICD11
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*Źródło

orphanet